Natural History and Biospecimen Repository for Dystonia; Comprehensive Rating Tools for Cervical Dystonia; Validity & Reliability of Diagnostic Methods & Measures of Spasmodic Dysphonia

Informed consent version date: August 3, 2016

NCT01373424

You Are Being Asked to Be in a Research Study

What Is a Research Study?
The main purpose of research studies is to gain knowledge. This knowledge may be used to help others. Research studies are not intended to benefit you directly, though some might.

Do I Have to Do This?
No. Being in this study is entirely your choice. If you decide to join this study, you can change your mind later on and withdraw from the research study.

Taking part in a study is separate from medical care. The decision to join or not join the research study will not affect your status as a patient.

What Is This Document?
This form is an informed consent document. It will describe the study risks, procedures, and any costs to you.

This form is also a HIPAA Authorization document. It will describe how your health information will be used and by whom.

Signing this form indicates you are willing to take part in the study and allow your health information to be used.

What Should I Do Next?
1. Read this form, or have it read to you.
2. Make sure the study doctor or study staff explains the study to you.
3. Ask questions (e.g., time commitment, unfamiliar words, specific procedures, etc.)
4. If there will be medical treatment, know which parts are research and which are standard care.
5. Take time to consider this, and talk about it with your family and friends.

Emory University School of Medicine

Research Participant Consent and
HIPAA Authorization to Use or Disclose Health Information that Identifies You for a Research Study

Title: Dystonia Coalition Project 1: Natural History and Biorepository

Principal Investigator: H. A. Jinnah, MD, PhD

Primary Sponsors: National Institutes of Health (NIH), Office of Rare Diseases Research in the National Center for Advancing Translational Sciences (ORDR-NCATS), National Institute of Neurological Disorders and Stroke (NINDS)

Sites: Emory Clinic, Executive Park, Emory University Hospital, Wesley Woods, hotels and homes

Introduction

You are being asked to be in a medical research study. This form is designed to tell you everything you need to think about before you decide to consent (agree) to be in the study or not to be in the study. **It is entirely your choice. If you decide to take part, you can change your mind later on and withdraw from the research study.** The decision to join or not join the research study will not cause you to lose any medical benefits. If you decide not to take part in this study, your doctor will continue to treat you.

- Please carefully read this form or have it read to you
- Please listen to the study doctor or study staff explain the study to you
- Please ask questions about anything that is not clear
- Feel free to take home an unsigned copy of this form and take your time to think about it and talk it over with family or friends

If you agree to join this research study, you will receive a copy of this consent form with your signature and the date to keep. Do not sign this consent form unless you have had a chance to ask questions and get answers that make sense to you. Nothing in this form can make you give up any legal rights. By signing this form you will not give up any legal rights.

Purpose

This study aims to create a “repository” or an organized collection used for future research on dystonia and other diseases. The repository will store biospecimens, videos, and clinical information. Biospecimens include things like blood, DNA, and cell lines from your body. Emory and other research centers will work together to put in information, videos, and samples from about ten thousand adults. These adults include people who have dystonia or related disorders. Researchers around the world will be able to use information, videos, and samples from all participants to do research that may lead to new tests and treatments. Your videos may be used to create teaching programs that will help medical professionals administer neurological exams or use specific rating scales.

Being in a research study does not take the place of routine physical exams or visits to your own doctor. You should not rely on this study to diagnose or treat medical problems.

A description of this clinical study will be available on http://www.ClinicalTrials.gov. This Web site will not include information that can identify you. At most the Web site will include a summary of the results. You may search this Web site at any time.

Procedures

You will be asked to come to the clinic on a convenient day for you or you may come on the same day as your regular scheduled clinic visit at Emory. Below are the activities we will ask you to do for the study and the time we expect them to take.

- We will look at your medical records.
- You will complete some questionnaires/interviews about your medical history, family history, thinking ability, mood, pain, activities of daily living, and quality of life. (15 to 45 minutes)
- You will have a neurological exam that will be vido recorded. For this exam you will do simple movements (like blink your eyes, turn your head to right and left, hold arms in front of you, walk up and down the hallway, etc…). This video will be used for this study and will be shared with other research teams studying dystonia and other movement disorders. (10 to 30 minutes)
- We will use a needle to draw about 2 tablespoons of blood from a vein in your arm. (5 to10 minutes)
- We will store your blood sample for use in this study and for future research studies, including genetic (DNA) tests.
- If you are participating in another study, the Dystonia Coalition's Blepharospasm Tools study (RDCNR#6304), the information collected in that study will be connected to the data collected for this study.
- We may contact you in the future by telephone, mail, or e-mail to ask for more information about your health or medical/family history for this study.
- We would like to be able to contact you in the future by telephone, mail, or email to ask if you are willing to participate in other, new research studies.

How long will I be in the study?

Each study visit will take 1 to 1.5 hours, depending on the questionnaires you are asked to complete. If your symptoms started 3 years ago or less, we will ask you to return annually for a follow-up visit. Once you have had symptoms for over 3 years, but less than 7, we will ask that you return once every 2 years for a follow-up visit. And once you have had symptoms for 7 years or more, we will ask that you return once every 4 years for a follow-up visit. These follow-up visits will include the same procedures as those listed above. A blood sample will only be requested at the follow-up visits if enough blood was not obtained during the first visit. You may decline to participate in any of the follow-up visits.

Any samples, videos, or clinical information will be used for many years to come. Your samples and data may be used for studies currently in progress or for studies that might occur in the future. The details, results, and implications of the studies that may use your samples and/or data are currently unknown

How will my samples and information be used?

All specimens, videos, and information will be stored in secure databases and specimen banks for future studies. Below are the various places where these items will be stored and what will be stored at each place.

1. Emory University
    a. All the data from the questionnaires (including identifiers like name, address, etc…)

b. DNA (genetic material from your blood cells)
c. Video(s), which have images of your face

2. Dystonia Coalition data center, currently at Washington University in St. Louis
   a. All the data from the questionnaires (including identifiers like name, address, etc…)
   b. DNA
   c. Video(s), which have images of your face

3. National Institute of Neurological Diseases and Stroke Repository (NINDS BR), a national, public resource, currently at the Coriell Institute for Medical Research (Coriell is funded by the NINDS to store, process, and distribute biospecimens, like DNA., and their associated data)
   a. De-identified information
   b. Year of Birth
   c. Blood Sample, including DNA

4. Office of Rare Diseases Research Data Management and Coordination Center (ORDR DMCC), currently located at the University of South Florida and a Federal data repository
   a. De-identified information
   b. Video(s), which have images of your face

The sponsor of this study will store your samples and information for an indefinite period of time. A code will be used to link your samples to your clinical information, your answers to the questionnaires, your neurological exam, your video, and any other records you release to the researchers. They will make your coded samples and information available to researchers at hospitals, universities, and commercial organizations. The samples could be used for research in any type of disease and other genetic factors, not just dystonia or neurologic diseases.

Your name and other identifying information will remain confidential, but people who know you may recognize your face if it is seen on the video.

The results of this study will not be used to change your treatment course, nor will they be recorded in your medical records. Because this is research and not a clinical exam, you will not receive any results from your individual testing and your study data will not be used to make treatment decisions. You will not receive a copy of your videotape(s). Research results will not be given to you or your physician. You will not receive any individual information regarding genetic studies which may be conducted on your blood sample. If there is a paper published using the data collected in this study, we will try to make the reference available on the Dystonia Coalition website: www.rarediseasesnetwork.org/dystonia. We will also try to make it available through Pubmed: http://www.ncbi.nlm.nih.gov/sites/entrez.

The DNA (genetic material) and white blood cells from your blood sample will be isolated and stored for future studies. These may include genetic testing. Permanent cell lines may be created from your sample. Cell lines are produced by growing blood cells in a laboratory and allow us to have a source of the DNA without having to redraw your blood. These blood cells can be stored for decades or more. The researchers will not (and are not allowed to) use your samples for cloning a human being. Your cells may be mixed with other human cells, mixed with animal cells, or grown in lab animals like mice. Your cells may be used to develop treatments for a variety fo diseases and conditions.

We may use the cells taken from your blood or skin to create a type of cell known as a pluripotent cell. This type of cell can be used to create different types of tissue, including neurons, muscle cells and other types of cells. Your cells might be used in research involving genetic alteration of the cells.

If researchers want to use your samples and associated data stored at the NINDS Repository, currently at Coriell, they must provide a Statement of Research Intent and an Assurance Form, signed by their institution, assuring appropriate use of the samples. In all other cases, when researchers want to use the samples, videos, or information collected in this study for research or creating training programs, they must get approval from their local Institutional Review Board. Then they may contact the ORDR DMCC, or researchers at the Dystonia Coalition data center or Emory University for permission.

It is possible that your sample may be used in a genetic study and the information from this study may be made freely available in a public, unrestricted database that anyone can use. For example, the public database could include information on hundreds of thousands of genetic variations in your DNA code, as well as your ethnic group, sex, and whether you have dystonia or not. This public information will not be labeled with your name or other information that could be used to easily identify you. However, it is possible that this information, when combined with information from other public sources could be used to identify you, though we believe it is unlikely this will happen.

How is my Genetic Information Protected? What are the Risks?

The Genetic Information Nondiscrimination Act (GINA) is a federal law that generally makes it illegal for health insurance companies, group health plans, and most employers to discriminate against you based on your genetic information. This law generally will protect you in the following ways:

- Health insurance companies and group health plans may not request your genetic information that we get from this research.
- Health insurance companies and group health plans may not use your genetic information when making decisions regarding your eligibility or premiums.
- Employers with 15 or more employees may not use your genetic information that we get from this research when making a decision to hire, promote, or fire you or when setting the terms of your employment.

Be aware that GINA does **not** protect you against genetic discrimination by companies that sell life insurance, disability insurance, or long-term care insurance, and does not apply to employers with less than 15 employees.

In addition to GINA, the State of Georgia has laws that prohibit insurers from using genetic testing information for any non-treatment purpose. However, like GINA, this state law protection has exclusions: life insurance policies, disability income policies, accidental death or dismemberment policies, medicare supplement policies, long-term care insurance policies, credit insurance policies, specified disease policies, hospital indemnity policies, blanket accident and sickness policies, franchise policies issued on an insurance policy written as a part of workers' compensation equivalent coverage, or other similar limited accident and sickness policies.

Privilege

In the State of Georgia, your genetic information has special legal protections called "privilege," which means that the information cannot be used as evidence in a court. By signing this form and allowing us to use and disclose your genetic information for the purposes described in this consent, you waive any privilege with regard to that genetic information, meaning that the information loses this legal protection.

Risks and Discomforts

You may have some discomfort, bruising, and/or bleeding at the site of the blood draw. You may feel dizzy or faint when your blood is drawn. There is also a slight risk of infection at the site of the blood draw. Some of the questions in the questionnaires may make you feel uncomfortable. You have the right to skip

any question you do not wish to answer. The videotaping of the examination may make you feel uncomfortable. Completing the study may be somewhat tiring, but you may quit at any time.

The video of your voice box may be uncomfortable. To lessen your discomfort, a nasal decongestant will be sprayed through your nasal cavities to open the nasal passages a few minutes before the procedure. There is also a small risk of minor nose bleeding.

There is a small risk of loss of confidentiality. A number of procedures are in place to protect participant confidentiality. Every participant enrolled in the study will receive a unique code number. All samples, sample containers, and videos will be labeled with this code number, and the code will be shared only with a limited number of researchers. Your neurological exam video and the video of your voice exercises have images of your face and will be shared with approved researchers. Because the image of your face is on the video(s), it is difficult to ensure total confidentiality once training videos are created or the videos are distributed for research. It is possible you could be identified by someone viewing these videos.

There is a risk that someone could use the information from the sample you submitted, via DNA, to identify you if it were matched with another DNA sample provided by you. However, any user of the sample must agree to not use it for that purpose, and the risk, while real, is small. There may be other risks that are currently unknown. For instance, in the future it may be possible to use the donated samples to determine genetic factors responsible for medical problems. If this information was disclosed it could possibly affect your ability to get insurance or a job. It could also affect your immediate family members this way. You may wish to discuss your participation in this study with your family members.

New Information

It is possible that the researchers will learn something new during the study about the risks of being in it. If this happens, they will tell you, so you can decide if you want to continue to be in this study. You may be asked to sign a new consent form that includes the new information if you decide to stay in the study.

Benefits

This study is not designed to benefit you directly. It is not a treatment study. This study is designed to help researchers learn more about dystonia and other diseases. The study results may be used to help other patients in the future.

Financial Issues

You will not be offered payment for being in this study.

The analysis of your samples and clinical information may contribute to the creation of new tests, drugs or other things that may be commercially valuable to the sponsor. There are no plans in place to share financial gains with you if this research leads to marketing new tests or treatments.

Other options

You are free to choose not to participate in this study.

Authorization to Use and Disclose Protected Health Information

Emory University and the Dystonia Coalition data center, currently at Washington University in St. Louis, will keep any research records we produce private to the extent we are required to do so by law. A study number rather than your name will be used on study records wherever possible. Your name and other facts that might point to you will not appear when we present this study or publish its results. However, your video will be used for research and teaching programs and does contain images of your face.

The privacy of your health information is important to us, especially if it can identify who you are. We call that your "protected health information" or "PHI." To protect your PHI, we will follow the federal and state privacy laws, including the Health Insurance Portability and Accountability Act and regulations (HIPAA). We refer to all of these laws as the "Privacy Rules". Here we let you know how we will use and disclose your PHI for the main study and for any optional activities in which you may choose to participate.

Main study

PHI that Will be Used/Disclosed:
The PHI that we will use or share for the main research study includes:

- All of the information in your medical record, including your medical history and present/past medications.
- Results of exams, procedures, questionnaires, and tests you have before and during the study.
- Video recordings of those exams, which will include images of your face
- Laboratory test results.
- Your name, initials, address, all elements of dates (like your date of birth or visit date), telephone, fax, cell phone numbers, email, URL, and medical record numbers.
- Date of birth, including year, will be used for everyone in the study, including people over 89 years of age.

Purposes for Which Your PHI Will be Used/Disclosed:
We will use and share your PHI for the conduct and oversight of the research study and for the conduct and oversight of this repository and of future studies that are covered by this consent and authorization. We will use and share your PHI to provide you with study related treatment and for payment for such treatment. We will also use and share your PHI to conduct normal business operations. We may share your PHI with other people and places that help us conduct or carry out the study, such as laboratories, data management centers, data monitors, contract research organizations, Institutional Review Boards (IRBs) and other study sites. If you leave the study, we may use your PHI to determine your health, vital status or contact information

Use and Disclosure of Your Information That is Required by Law:
We will use and disclose your PHI when we are required to do so by law. This includes laws that require us to report child abuse or abuse of elderly or disabled adults. We will also comply with legal requests or orders that require us to disclose your PHI. These include subpoenas or court orders.

Authorization to Use PHI is Required to Participate:
By signing this form, you give us permission to use and share your PHI as described in this document. You do not have to sign this form to authorize the use and disclosure of your PHI. If you do not sign this form, then you may not participate in the research study.

People Who will Use/Disclose Your PHI:
The following people and groups will use and disclose your PHI in connection with the research study:

- The Principal Investigator and the research staff will use and disclose your PHI to conduct the study
- Emory may use and disclose your PHI to run normal business operations.
- The Principal Investigator and research staff will share your PHI with other people and groups to help conduct the study or to provide oversight for the study.
- The National Institutes of Health (NIH), National Institute of Neurological Disorders and Stroke (NINDS), Office of Rare Disease Research (ORDR) at the National Center for Advancing

Translational Sciences (NCATS) are the Sponsors of the study. The Sponsors may use and disclose your PHI to make sure the research is done correctly and to collect and analyze the results of the research. The Sponsors may disclose your PHI to other people and groups like study monitors to help conduct the study or to provide oversight for the study.

- Future researchers
- The following people and groups will use your PHI to make sure the research is done correctly and safely:
    - Emory offices that are part of the Human Research Participant Protection Program and those that are involved in study administration and billing. These include the Emory IRB, the Emory Research and Healthcare Compliance Offices, and the Emory Office for Clinical Research.
    - Washington University in St. Louis Institutional Review Board
    - Government agencies that regulate the research including: Office for Human Research Protections
    - Public health agencies.
    - Research monitors and reviewers.
    - Accreditation agencies.
    - Medical Review Officers
- Sometimes a Principal Investigator or other researcher moves to a different institution. If this happens, your PHI may be shared with that new institution and their oversight offices. PHI will be shared securely and under a legal agreement to ensure it continues to be used under the terms of this consent and HIPAA authorization.

Training video activity & future contact for other studies (optional):

PHI That Will be Used/Disclosed for optional activities:
The PHI that we will use and/or disclose (share) for the optional activities includes: the same items that are listed above for the main study.

Purposes for which your PHI will be Used/Disclosed for optional activities:
We will use and disclose your PHI for the conduct and oversight of the optional activities.

Authorization for This Use of PHI is Required to Participate in Optional Activities:
You do not have to authorize the use and disclosure of your PHI for these optional activities. If you do not authorize the use and disclosure of your PHI for the optional activities, then you may not participate in the optional activities. You can still be in the main research study even if you don't participate in the optional activities.

People Who Will Use/Disclose Your PHI for Optional Study:
The same people and groups who will use and disclose your PHI for the main study will also do so in connection with the optional research study

Other Items You Should Know:
HIPAA only applies to people or organizations that are health care providers, health care payers or healthcare clearinghouses. HIPAA may not apply to all Information Users. If HIPAA does not apply to an Information User, then that User does not have to follow HIPAA requirements when it uses or discloses your health information.

You do not have to sign this authorization form, but if you do not, you may not take part in the research study. Your decision whether or not to participate in this study will not affect your medical care.

You generally will not have access to your PHI related to this research study until the study is over. When the study is done, at your request, you may generally have access to any of your study-related PHI that is also part of the PHI your health care providers use to make decisions about your care. Your doctor may be able to access this PHI before the study is over if it is needed for your treatment.

If the PHI is removed from your health information, then the leftover information would not be covered by HIPAA, and it may be used or passed along to other persons or groups, and/or for other purposes.

Expiration Date: This authorization does not have an expiration date.

Withdrawal from the Study or Revoking your Authorization

You will be given a signed copy of this consent form for your records. If your ability to consent for yourself changes, you or your legal representative may be asked to re-consent prior to continued participation. The research investigators may withdraw you from the study without your consent if considered appropriate.

You do not have to sign this form. If you do not, you will not be able to participate in this research study. If you sign this consent and authorization, you may change your mind at any time and decide you do not want to participate anymore. If you want to revoke (take back) this authorization you must write to: Dr. H. A. Jinnah, 101 Woodruff Circle, 6305 Woodruff Memorial Building, Atlanta, GA 30322. Not signing this form, revoking your authorization and consent, or withdrawing from the study will not affect your medical treatment.

If you revoke your authorization, the researchers will not collect any more health information that identifies you, but they may use or disclose identifiable information that you already gave them in order to notify any of the other Information Users that you have taken back your authorization; to maintain the integrity or reliability of the Research Study; and to comply with any law that they are required to obey. You have the right to withdraw from this research project at any time. If possible, any samples you have contributed will be discarded if you request this; however, because of the sample masking, we may not always be able to identify which samples were donated by you. We will remove your name, address and other information that may identify you, including your videotaped examination(s) from the research record. Although some of the information you have provided may remain in the research record, no one will ever be able to trace the source of information and know that it came from you. If your video has already been included in a teaching program, we will attempt to have it removed from future versions, but this may not be possible.

In Case of Injury

If you get ill or injured from being in this study, Emory will arrange for you to have urgent health care. If you believe you have been injured by this research, you should contact Dr. Jinnah (404-727-9107*).* Here we explain who would pay for this health care*:*

Would Emory Pay? Emory has not set aside any funds to pay for urgent health care. Also, Emory has not set aside any funds to pay you if you become ill or injured from being in this study. The only exception to this policy is if it is proven that the negligence of an Emory employee directly caused your injury or illness. "Negligence" means the failure to follow a standard duty of care.

Would the Study Sponsors (NIH/NINDS/ORDR/Allergan/Dystonia Study Group/National Spasmodic Dysphonia Association) Pay? There is no program for compensation through the National Institute of Neurological Disorders and Stroke (NINDS)/National Institutes of Health (NIH) or any of the other sponsors.

Costs

There will be no costs to you for taking part in this study, except maybe for the cost of travel to Emory. If you are a patient at Emory, your clinic or hospital visit is not part of the study. You or your health plan will be responsible for the cost of your regular medical care.

Questions

Contact the research coordinators (Adam Kassem) at 404-727-3381 or akassem@emory.edu or the Principal Investigator (Dr. H. A. Jinnah) at 404-727-9107 or hjinnah@emory.edu:

- if you have any questions about this study or your part in it,
- if you feel you have had a research-related injury, or
- if you have questions, concerns or complaints about the research.

If you have questions about your rights as a research participant or if you have questions, concerns or complaints about the research, you may contact the Emory University Institutional Review Board at 404-712-0720, 877-503-9797, or irb@emory.edu.

Consent

TO BE FILLED OUT BY SUBJECT ONLY

There are two optional activities associated with this study. Please initial yes if you agree and no if you do not.

Yes	No	
_____	_____	The researchers are allowed to create training videos using my video(s). I am aware that on a training tape my face and body will be visible and there is a possibility I could be recognized.
_____	_____	The researchers are allowed to contact me by telephone, mail, or e-mail for future follow-up studies

Please **print** your name, **sign**, and **date** below if you agree to be in the study. By signing this consent and authorization form, you will not give up any of your legal rights. We will give you a copy of the signed form to keep. You have read this consent and authorization form (or it has been read to you). All your questions about the study and your part in it have been answered. You freely consent to be in this research study.

_____
Name of Participant

_____ _____
Signature of Participant Date/Time

TO BE FILLED OUT BY STUDY TEAM ONLY

_____
Name of Person Conducting Informed Consent Discussion

_____ _____
Signature of Person Conducting Informed Consent Discussion Date